SPARK Gene List

Updated September 2022

SPARK

Autism Gene Discovery

Genetic changes are one of many causes of autism. Scientists have identified **over 200 genes** and segments of chromosomes, known as copy number variants (CNVs), that are related to autism. But there is still so much to learn.

Scientists predict that **several hundred more genes related to autism have yet to be found**. One of SPARK's main goals is to find them.

The more people who participate in SPARK, the faster we can find these genetic changes. If you or your family member has autism, **your DNA may contain important clues** that can help us to better understand autism genetics.

What are genes, chromosomes and genetic differences?

Genes

Genes are made up of DNA. They provide instructions for making the proteins that our cells and body need in order to function.

We all have the same genes. For example, we all have the gene called, CHD8.

Chromosomes

Genes are located on chromosomes.

Everybody has one set of chromosomes from their mother and one set from their father.

Genetic differences

Except for identical twins, no two people have the same genetic make-up. Everyone has genetic differences that make them unique.

Genetic differences can vary in their size, but a small change can still have a big impact:

- Some people, such as those with Down syndrome, have extra copies of entire chromosomes.
- Some people have chromosomes with regions that have been deleted, duplicated or rearranged. These changes, which scientists call copy number variants, often include multiple genes.
- Some people have smaller genetic differences within single genes.

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The SPARK gene list contains 162 single genes (blue), 43 copy number variants (orange) and 5 chromosomal differences (green) that are known to be associated with autism. More information about each autism-linked gene or CNV, along with its associated symptoms, is available **here**.

Copy Number Variants

- 1q21.1 del
- 1q21.1 dup
- 2p16.3 del
- 2q37.3 del
- 3q29 del
- 3q29 dup
- 5p- del
- 5q35 del
- 5q35 dup
- 6q16 del
- 7q11.23 del
- 7q11.23 distal del
- 7q11.23 dup
- 8p23.1 dup
- 9q34 del
- 9q34 dup
- 15q11.2q13.1 del
- 15q11.2q13.1 dup
- 15q13.3 del
- 15q15 del
- 15q24A_C del
- 16p11.2 del
- 16p11.2 distal del
- 16p11.2 dup
- 16p12.2 del
- 16p13.11 del
- 16p13.3 del
- 17p11.2 del
- 17p11.2 dup
- 17p13.3 del
- 17p13.3 dup
- 17q11.2 del
- 17q11.2 dup
- 17q12 del
- 17q12 dup
- 17q21.3 del
- 17q21.3 dup
- 22q11.2 del
- 22q11.2 dup
- 22q11.2 central del
- 22q11.2 central dup
- 22q13.3 del
- Xq28 dup

Chromosomal Differences

- Trisomy 21 (Down syndrome)
- XO (Turner syndrome)
- XXY (Klinefelter syndrome)
- XXYY
- XXXY

Single Genes

- ACTB
- ADNP
- ADSL*
- AFF2
- AHDC1
- ALDH5A1*
- ANK2
- ANK3*
- ANKRD11
- ARHGEF9
- ARID1B
- ARX
- ASH1L
- ASXL3
- ATRX
- AUTS2
- BCKDK*
- BCL11A
- BRAF
- BRSK2
- CACNA1C
- CAPRIN1
- CASK
- CASZ1
- CBL
- CDKL5
- CHAMP1
- CHD2
- CHD3
- CHD7
- CHD8
- CIC
- CNOT3
- CREBBP
- CSDE1
- CTCF
- CTNNB1
- CUL3
- DDX3X
- DEAF1*
- DHCR7*
- DLG4
- DMPK
- DNMT3A
- DSCAM
- DYNC1H1
- DYRK1A
- EBF3
- EHMT1
- EIF3F*
- EP300
- FMR1
- FOXG1
- FOXP1
- GIGYF1
- GRIN1*
- GRIN2A*
- GRIN2B
- HIVEP2
- HNRNPH2*
- HNRNPU
- HRAS
- IQSEC2
- IRF2BPL
- KANSL1
- KCNB1
- KCNQ3*
- KDM3B
- KDM6B
- KMT2A
- KMT2C
- KMT5B
- KRAS
- LZTR1
- MAGEL2
- MAP2K1
- MAP2K2
- MBD5
- MBOAT7*
- MECP2
- MED13
- MED13L
- MEF2C
- MEIS2
- MTOR*
- MYT1L
- NAA15
- NBEA
- NCKAP1
- NEXMIF
- NF1
- NIPBL
- NLGN2
- NLGN3
- NLGN4X
- NR4A2
- NRAS
- NRXN1
- NRXN2
- NSD1
- PACS1*
- PCDH19
- PHF21A
- PHF3
- PHIP
- POGZ
- POMGNT1*
- PPP1CB
- PPP2R5D*
- PSMD12
- PTCHD1
- PTEN
- PTPN11
- RAF1
- RAI1
- RALGAPB
- RELN*
- RERE
- RFX3
- RIT1
- RORB
- SCN1A
- SCN2A
- SCN8A
- SETBP1
- SETD2
- SETD5
- SHANK2
- SHANK3
- SHOC2
- SIN3A
- SLC6A1
- SLC9A6
- SMARCC2
- SON
- SOS1
- SOS2
- SOX5
- SPAST
- SRCAP
- STXBP1
- SYNGAP1
- TANC2
- TAOK1
- TBCK*
- TBR1
- TCF4
- TCF20
- TLK2
- TRIO
- TRIP12
- TSC1
- TSC2
- UBE3A
- UPF3B
- VPS13B*
- WAC
- WDFY3
- YY1*
- ZBTB20
- ZNF292
- ZNF462

**Only certain variants in these genes are returned.*

How does a gene, CNV, or chromosomal difference make it onto the list?

We include genes, CNVs, and chromosomal differences that have strong and consistent evidence that they are associated with autism.

We update the list four times a year. New genes, CNVs, and chromosomal differences are approved by the SPARK medical genetics committee.

What is the SPARK medical genetics committee?

The members of the committee are experts in the study of autism genetics.

The committee meets four times a year with SPARK's principal investigator, Dr. Wendy Chung, and SPARK's scientific director, Dr. Pamela Feliciano, to discuss the genetic findings from the SPARK study as well as updates to the SPARK gene list.

You can learn more about the members of the committee in this video.

How does SPARK inform participants of genetic results?

When we find that a participant with autism has a genetic change in one of the genes or CNVs on our list, we share those results with you as soon as we can, free of charge.

Participants may decide if they want to hear about their results through their own medical provider or a SPARK-provided genetic counselor.

It may take months or years for a participant to get a result. That's because SPARK's analyses are complex, and they take into account the latest advances in autism genetics.

At this time, we are finding genetic changes related to autism in about 10 percent of families enrolled in SPARK.

To check the status of your sample(s),
log in to your SPARK dashboard.

Contact us at infoSPARKforAutism.org
if you have any questions.